



## PYGL gene

glycogen phosphorylase L

### Normal Function

The *PYGL* gene provides instructions for making an enzyme called liver glycogen phosphorylase. This enzyme breaks down a complex sugar called glycogen. Liver glycogen phosphorylase is one of three related enzymes that break down glycogen in cells; the other glycogen phosphorylases are found in the brain and in muscles. Liver glycogen phosphorylase is found only in liver cells, where it breaks down glycogen into a type of sugar called glucose-1-phosphate. Additional steps convert glucose-1-phosphate into glucose, a simple sugar that is the main energy source for most cells in the body.

### Health Conditions Related to Genetic Changes

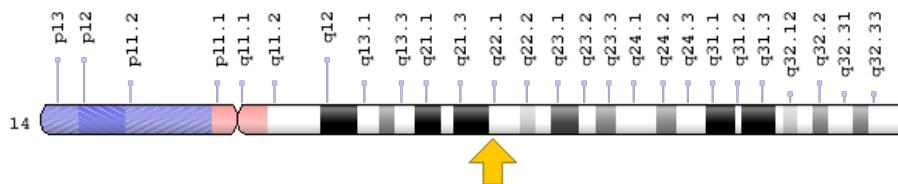
#### glycogen storage disease type VI

At least 17 mutations in the *PYGL* gene have been found to cause glycogen storage disease type VI (GSDVI). Most mutations change single protein building blocks (amino acids) in liver glycogen phosphorylase, affecting the normal function of the enzyme. In the Old Order Mennonite population, a common mutation (written as 1620+1G>A) disrupts the way the *PYGL* gene's instructions are used to make the enzyme. A defective liver glycogen phosphorylase enzyme impairs the normal breakdown of glycogen. As a result, liver cells cannot use glycogen for energy, so liver function becomes impaired. A lack of glycogen breakdown within liver cells leads to the major features of GSDVI.

## Chromosomal Location

Cytogenetic Location: 14q22.1, which is the long (q) arm of chromosome 14 at position 22.1

Molecular Location: base pairs 50,905,217 to 50,944,530 on chromosome 14 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- glycogen phosphorylase, liver form
- GSD6
- phosphorylase, glycogen, liver
- PYGL\_HUMAN

## Additional Information & Resources

### Educational Resources

- Biochemistry (fifth edition, 2002): Glucose Regulation of Liver Glycogen Metabolism  
<https://www.ncbi.nlm.nih.gov/books/NBK22444/?rendertype=figure&id=A2963>
- Biochemistry (fifth edition, 2002): Glycogen Metabolism in the Liver Regulates the Blood-Glucose Level  
<https://www.ncbi.nlm.nih.gov/books/NBK22444/#A2961>
- Biochemistry (fifth edition, 2002): Phosphorylase Catalyzes the Phosphorolytic Cleavage of Glycogen to Release Glucose 1-phosphate  
<https://www.ncbi.nlm.nih.gov/books/NBK22467/#A2918>

### GeneReviews

- Glycogen Storage Disease Type VI  
<https://www.ncbi.nlm.nih.gov/books/NBK5941>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28PYGL%5BTIAB%5D%29+OR+%28glycogen+phosphorylase+liver%5BALL%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- GLYCOGEN PHOSPHORYLASE, LIVER  
<http://omim.org/entry/613741>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=PYGL%5Bgene%5D>
- HGNC Gene Family: Glycogen phosphorylases  
<http://www.genenames.org/cgi-bin/genefamilies/set/437>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=9725](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9725)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/5836>
- UniProt  
<http://www.uniprot.org/uniprot/P06737>

## **Sources for This Summary**

- Beauchamp NJ, Taybert J, Champion MP, Layet V, Heinz-Erian P, Dalton A, Tanner MS, Pronicka E, Sharrard MJ. High frequency of missense mutations in glycogen storage disease type VI. *J Inherit Metab Dis.* 2007 Oct;30(5):722-34. Epub 2007 Aug 21.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17705025>
- Burwinkel B, Bakker HD, Herschkovitz E, Moses SW, Shin YS, Kilimann MW. Mutations in the liver glycogen phosphorylase gene (PYGL) underlying glycogenosis type VI. *Am J Hum Genet.* 1998 Apr;62(4):785-91.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/9529348>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1377030/>
- Chang S, Rosenberg MJ, Morton H, Francomano CA, Biesecker LG. Identification of a mutation in liver glycogen phosphorylase in glycogen storage disease type VI. *Hum Mol Genet.* 1998 May;7(5):865-70.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/9536091>
- GeneReview: Glycogen Storage Disease Type VI  
<https://www.ncbi.nlm.nih.gov/books/NBK5941>

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